

PX018432US_SEQ_1stOA_Amended.txt

<110> Samsung Electronics Co. Ltd

<120> A variant of HNF-1a gene having novel single nucleotide polymorphism and a variant protein encoded by the same

<130> YPL-0064

<140> 10/6663,857

<141> 2003-09-15

<160> 27

<170> KopatentIn 1.71

<210> 1

<211> 1896

<212> DNA

<213> Homo sapiens

<220>

<221> CDS

<222> (1)..(1893)

<223> amino acid sequence of HNF-1a

<400> 1

atg gtt tct aaa ctg agc cag ctg cag acg gag ctc ctg gcg gcc ctg
Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu
1 5 10 15

48

ctc gag tca ggg ctg agc aaa gag gca ctg atc cag gca ctg ggt gag
Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu
20 25 30

96

ccg ggg ccc tac ctc ctg gct gga gaa ggc ccc ctg gac aag ggg gag
Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu
35 40 45

144

tcc tgc ggc ggc ggt cga ggg gag ctg gct gag ctg ccc aat ggg ctg
Ser Cys Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
50 55 60

192

ggg gag act cgg ggc tcc gag gac gag acg gac gac gat ggg gaa gac
Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp
65 70 75 80

240

ttc acg cca ccc atc ctc aaa gag ctg gag aac ctc agc cct gag gag
Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu
85 90 95

288

gcg gcc cac cag aaa gcc gtg gtg gag acc ctt ctg cag gag gac ccg
Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro
100 105 110

336

tgg cgt gtg gcg aag atg gtc aag tcc tac ctg cag cag cac aac atc
Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile
115 120 125

384

cca cag cgg gag gtg gtc gat acc act ggc ctc aac cag tcc cac ctg

432

PX018432US_SEQ_1stOA_Amended.txt

Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu	130	135	140	
tcc caa cac ctc aac aag ggc act ccc atg aag acg cag aag cgg gcc				480
Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala	145	150	155	160
gcc ctg tac acc tgg tac gtc cgc aag cag cga gag gtg gcg cag cag				528
Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln	165	170	175	
ttc acc cat gca ggg cag gga ggg ctg att gaa gag ccc aca ggt gat				576
Phe Thr His Ala Gly Gln Gly Leu Ile Glu Glu Pro Thr Gly Asp	180	185	190	
gag cta cca acc aag aag ggg cgg agg aac cgt ttc aag tgg ggc cca				624
Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro	195	200	205	
gca tcc cag cag atc ctg ttc cag gcc tat gag agg cag aag aac cct				672
Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro	210	215	220	
agc aag gag gag cga gag acg cta gtg gag gag tgc aat agg gcg gaa				720
Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu	225	230	235	240
tgc atc cag aga ggg gtg tcc cca tca cag gca cag ggg ctg ggc tcc				768
Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser	245	250	255	
aac ctc gtc acg gag gtg cgt gtc tac aac tgg ttt gcc aac cgg cgc				816
Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg	260	265	270	
aaa gaa gaa gcc ttc cgg cac aag ctg gcc atg gac acg tac agc ggg				864
Lys Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly	275	280	285	
ccc ccc cca ggg cca ggc ccg gga cct gcg ctg ccc gct cac agc tcc				912
Pro Pro Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser	290	295	300	
cct ggc ctg cct cca cct gcc ctc tcc ccc agt aag gtc cac ggt gtg				960
Pro Gly Leu Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val	305	310	315	320
cgc tat gga cag cct gcg acc agt gag act gca gaa gta ccc tca agc				1008
Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser	325	330	335	
agc ggc ggt ccc tta gtg aca gtg tct aca ccc ctc cac caa gtg tcc				1056
Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser	340	345	350	
ccc acg ggc ctg gag ccc agc cac agc ctg ctg agt aca gaa gcc aag				1104
Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys	355	360	365	

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ctg gtc tca gca gct ggg ggc ccc ctc ccc cct gtc agc acc ctc aca Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr 370 375 380	1152
gca ctg cac agc ttg gag cag aca tcc cca ggc ctc aac cag cag ccc Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro 385 390 395 400	1200
cag aac ctc atc atg gcc tca ctt cct ggg gtc atg acc atc ggg cct Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro 405 410 415	1248
ggt gag cct gcc tcc ctg ggt cct acg ttc acc aac aca ggt gcc tcc Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser 420 425 430	1296
acc ctg gtc atc ggc ctg gcc tcc acg cag gca cag agt gtg ccg gtc Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val 435 440 445	1344
atc aac agc atg ggc agc agc ctg acc acc ctg cag ccc gtc cag ttc Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe 450 455 460	1392
tcc cag ccg ctg cac ccc tcc tac cag cag ccg ctc atg cca cct gtg Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val 465 470 475 480	1440
cag agc cat gtg acc cag aac ccc ttc atg gcc acc atg gct cag ctg Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu 485 490 495	1488
cag agc ccc cac gcc ctc tac agc cac aag ccc gag gtg gcc cag tac Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr 500 505 510	1536
acc cac acg ggc ctg ctc ccg cag act atg ctc atc acc gac acc acc Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr 515 520 525	1584
aac ctg agc gcc ctg gcc agc ctc acg ccc acc aag cag gtc ttc acc Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr 530 535 540	1632
tca gac act gag gcc tcc agt gag tcc ggg ctt cac acg ccg gca tct Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser 545 550 555 560	1680
cag gcc acc acc ctc cac atc ccc agc cag gac cct gcc ggc atc cag Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln 565 570 575	1728
cac ctg cag ccg cac cgg ctc agc gcc agc ccc aca gtg tcc tcc His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser 580 585 590	1776
agc agc ctg gtg ctg tac cag agc tca gac tcc agc aat ggc cag agc Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser 595 600 605	1824

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His	Leu	Leu	Pro	Ser	Asn	His	Ser	Val	Ile	Glu	Thr	Phe	Ile	Ser	Thr	
610						615				620						
cag	atg	gcc	tct	tcc	tcc	cag				taa						1896
Gln	Met	Ala	Ser	Ser	Ser	Gln										
625			630													
<210> 2																
<211> 631																
<212> PRT																
<213> Homo sapiens																
<400> 2																
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Leu	Glu	Ser	Gly	Leu	Ser	Lys	Glu	Ala	Leu	Ile	Gln	Ala	Leu	Gly	Glu	
				20					25				30			
Pro	Gly	Pro	Tyr	Leu	Leu	Ala	Gly	Glu	Gly	Pro	Leu	Asp	Lys	Gly	Glu	
				35			40				45					
Ser	Cys	Gly	Gly	Gly	Arg	Gly	Glu	Leu	Ala	Glu	Leu	Pro	Asn	Gly	Leu	
					50		55			60						
Gly	Glu	Thr	Arg	Gly	Ser	Glu	Asp	Glu	Thr	Asp	Asp	Asp	Gly	Glu	Asp	
				65		70			75				80			
Phe	Thr	Pro	Pro	Ile	Leu	Lys	Glu	Leu	Glu	Asn	Leu	Ser	Pro	Glu	Glu	
				85				90				95				
Ala	Ala	His	Gln	Lys	Ala	Val	Val	Glu	Thr	Leu	Leu	Gln	Glu	Asp	Pro	
				100				105				110				
Trp	Arg	Val	Ala	Lys	Met	Val	Lys	Ser	Tyr	Leu	Gln	Gln	His	Asn	Ile	
					115		120			125						
Pro	Gln	Arg	Glu	Val	Val	Asp	Thr	Thr	Gly	Leu	Asn	Gln	Ser	His	Leu	
				130		135			140							
Ser	Gln	His	Leu	Asn	Lys	Gly	Thr	Pro	Met	Lys	Thr	Gln	Lys	Arg	Ala	
				145		150			155				160			
Ala	Leu	Tyr	Thr	Trp	Tyr	Val	Arg	Lys	Gln	Arg	Glu	Val	Ala	Gln	Gln	
				165			170			175						
Phe	Thr	His	Ala	Gly	Gln	Gly	Leu	Ile	Glu	Glu	Pro	Thr	Gly	Asp		
				180			185			190						
Glu	Leu	Pro	Thr	Lys	Lys	Gly	Arg	Arg	Asn	Arg	Phe	Lys	Trp	Gly	Pro	
				195			200			205						
Ala	Ser	Gln	Gln	Ile	Leu	Phe	Gln	Ala	Tyr	Glu	Arg	Gln	Lys	Asn	Pro	
				210		215			220							
Ser	Lys	Glu	Glu	Arg	Glu	Thr	Leu	Val	Glu	Glu	Cys	Asn	Arg	Ala	Glu	

PX018432US_SEQ_1stOA_Amended.txt

225	230	235	240
Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser			
245	250	255	
Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg			
260	265	270	
Lys Glu Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly			
275	280	285	
Pro Pro Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser			
290	295	300	
Pro Gly Leu Pro Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val			
305	310	315	320
Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser			
325	330	335	
Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser			
340	345	350	
Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys			
355	360	365	
Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr			
370	375	380	
Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro			
385	390	395	400
Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro			
405	410	415	
Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser			
420	425	430	
Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val			
435	440	445	
Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe			
450	455	460	
Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val			
465	470	475	480
Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu			
485	490	495	
Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr			
500	505	510	
Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr			
515	520	525	
Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr			
530	535	540	

PX018432US_SEQ_1stOA_Amended.txt

Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser
545 550 555 560

Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln
565 570 575

His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser
580 585 590

Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser
595 600 605

His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
610 615 620

Gln Met Ala Ser Ser Gln
625 630

<210> 3
<211> 93
<212> DNA
<213> Homo sapiens

<400> 3
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ccccgggctc aggaggctgc tctgctcccc cag 93

<210> 4
<211> 41
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying promoter of MODY3 gene

<400> 4
taatacgtact cactataggg tggccgtgag catcctctgc c 41

<210> 5
<211> 39
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying promoter of MODY3 gene

<400> 5
gtaaccctca ctaaaggac gtgggttgcg tttgcctgc 39

<210> 6
<211> 40
<212> DNA

PX018432US_SEQ_1stOA_Amended.txt

<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 1 of MODY3 gene

<400> 6
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<210> 7
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> antisense primer for amplifying exon 1 of MODY3 gene

<400> 7
gtaaccctca ctaaaggag ggctcgtag gagctgaggg 40

<210> 8
<211> 42
<212> DNA
<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 2 of MODY3 gene

<400> 8
taatacgaact cactataggg cccttgctga gcagatcccg tc 42

<210> 9
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> antisense primer for amplifying exon 2 of MODY3 gene

<400> 9
gtaaccctca ctaaaggag gatatggtaa gcttccagcc 40

<210> 10
<211> 40
<212> DNA
<213> Artificial Sequence

<220>

<223> sense primer for amplifying exon 3 of MODY3 gene

<400> 10

taatacgact cactataggg gcaaggtcag ggaaatggac

40

<210> 11
 <211> 42
 <212> DNA
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<220>
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42

<210> 12
 <211> 43
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 <213> Artificial Sequence

<220>
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<400> 12
 taatacgact cactataggg ggctcatggg tggctatttc tgc

43

<210> 13
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 4 of MODY3 gene

<400> 13
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42

<210> 14
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 5 of MODY3 gene

<400> 14
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42

<210> 15
 <211> 42
 <212> DNA
 <213> Artificial Sequence

PX018432US_SEQ_1stOA_Amended.txt

<220> antisense primer for amplifying exon 5 of MODY3 gene

<400> 15
gtaaccctca ctaaaggat acaagcaagg acactcacca gc 42

<210> 16
<211> 41
<212> DNA
<213> Artificial Sequence

<220> sense primer for amplifying exon 6 of MODY3 gene

<400> 16
taatacgaact cactataggg cccggacaca gcttggcttc c 41

<210> 17
<211> 42
<212> DNA
<213> Artificial Sequence

<220> antisense primer for amplifying exon 6 of MODY3 gene

<400> 17
gtaaccctca ctaaaggaa tccccaccag cttaccgatg ac 42

<210> 18
<211> 40
<212> DNA
<213> Artificial Sequence

<220> sense primer for amplifying exon 7 of MODY3 gene

<400> 18
taatacgaact cactataggg caggcctggc ctccacgcag 40

<210> 19
<211> 40
<212> DNA
<213> Artificial Sequence

<220> antisense primer for amplifying exon 7 of MODY3 gene

<400> 19
gtaaccctca ctaaaggag gggctctgca gctgagccat 40

PX018432US_SEQ_1stOA_Amended.txt

<210> 20
<211> 41
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 8 and 9 of MODY3 gene

<400> 20
taatacgaact cactataggg ggcccgatc acccacacgg g 41

<210> 21
<211> 40
<212> DNA
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<220>
<223> antisense primer for amplifying exon 8 and 9 of MODY3 gene

<400> 21
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<210> 22
<211> 41
<212> DNA
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<220>
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<400> 22
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<210> 23
<211> 41
<212> DNA
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<220>
<223> antisense primer for amplifying exon 10 of MODY3 gene

<400> 23
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<210> 24
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<213> Artificial Sequence

PX018432US_SEQ_1stOA_Amended.txt

<220>

<223> T7 promoter sequence

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20

<210> 25

<211> 19

<212> DNA

<213> Artificial Sequence

<220>

<223> T3 promoter sequence

<400> 25

gtaaccctca ctaaaaggga

19

<210> 26

<211> 540

<212> DNA

<213> Homo sapiens

<400> 26

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60

ccagccctct acagccacaa gcccggatgt gcccagtaca cccacacggg cctgctcccg

120

cagactatgc tcatcaccga caccaccaac ctgagcgccc tggccagcct cacgcccacc

180

aagcaggtaa ggtccaggcc tgctggccct ccctcggcct gtgacagagc ccctcacc

240

cacatcccccc gggctcagga ggctgctctg ctcccccagg tcttcacccctc agacactgag

300

gcctccagtg agtccggcgt tcacacgccc gcatctcagg ccaccacccct ccacatcccc

360

agccaggacc ctgcccggcat ccagcacctg cagccggccc accggctcag cgccagcccc

420

acaggtgaga ggcctggct ccacccctc cttactgtc cctgccccct tccatgttgg

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540

540

<210> 27

<211> 540

<212> DNA

<213> Homo sapiens

<400> 27

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60

ccagccctct acagccacaa gcccggatgt gcccagtaca cccacacggg cctgctcccg

120

cagactatgc tcatcaccga caccaccaac ctgagcgccc tggccagcct cacgcccacc

180

PX018432US_SEQ_1stOA_Amended.txt

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gcctccagtg agtccgggct tcacacgccg gcatctcagg ccaccacccct ccacgtcccc	360
agccaggacc ctgcccccat ccagcacctg cagccggccc accggctcag cgccagcccc	420
acaggtgaga ggccctggct ccacccctc ccttactgtc cctgccccct tccatgttgg	480
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